The Value of Nasal Bone & Cerebral Lateral Ventricle

In First and Second Trimester of Pregnancy

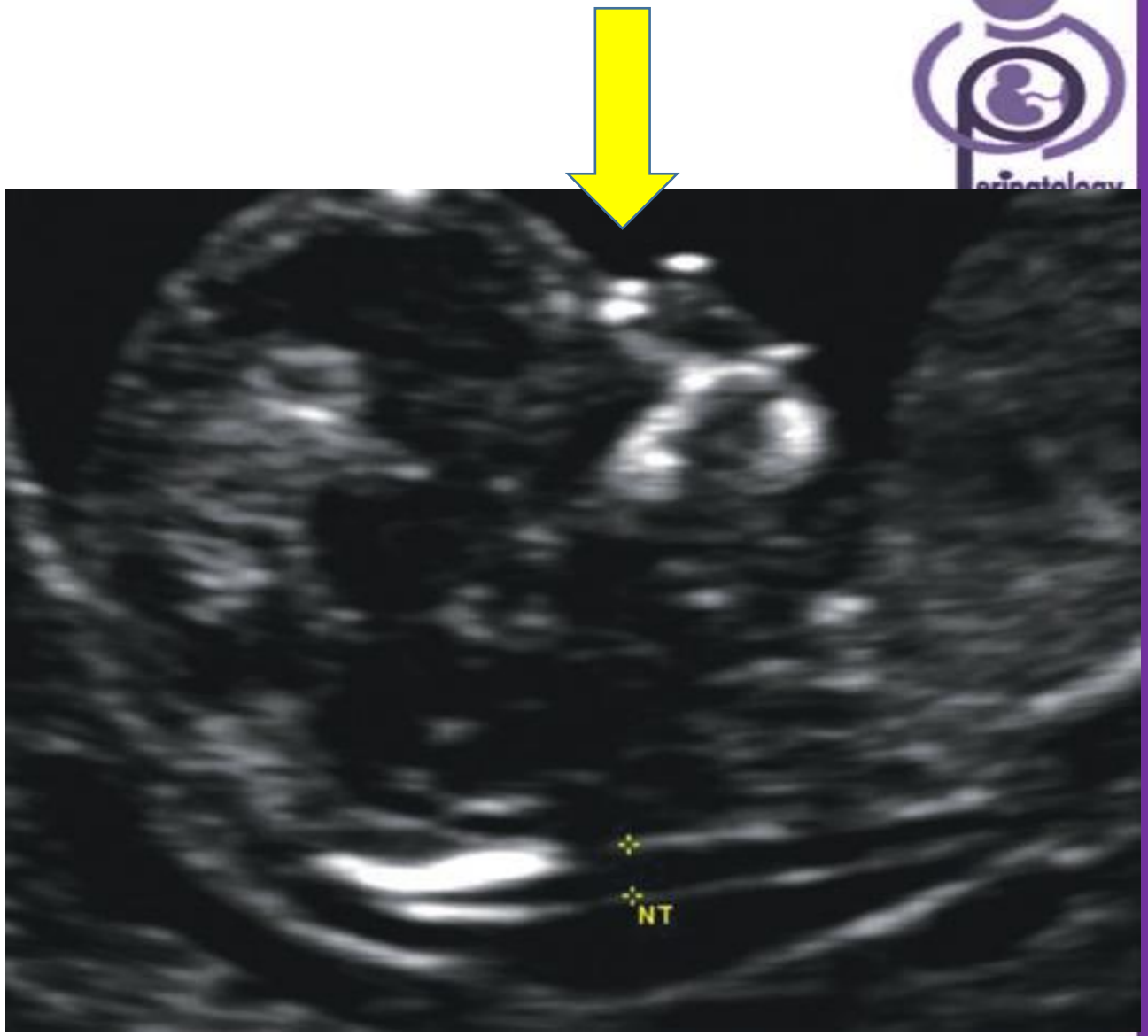
http://perinatalrc.sums.ac.ir

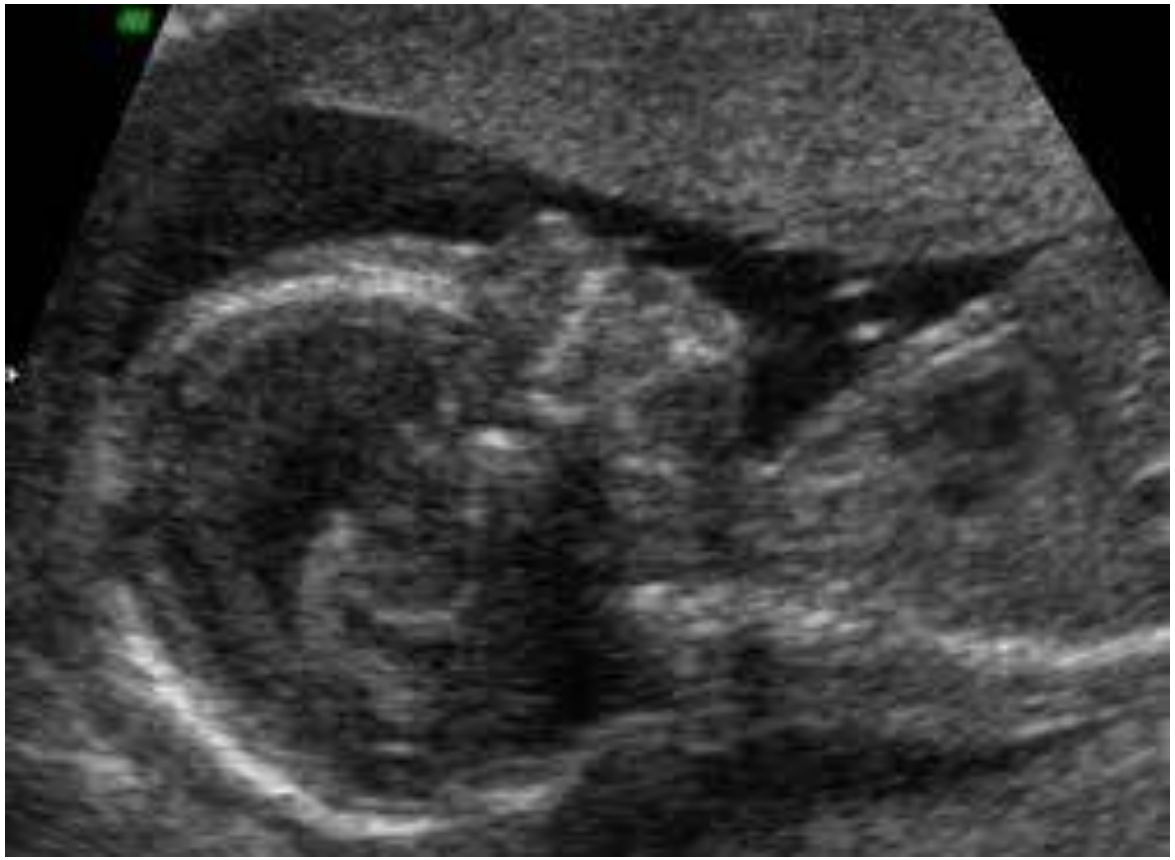
First trimester screening
&
Absent Nasal Bone

- The appearance of a small nose with a low nasal bridge in children and adults with Down syndrome initially led to the investigation of the fetal nasal bone as a marker for this condition.

- A ۲۰۰۳ study performed NT screening found that the nasal bone was absent in ۷۰% of fetuses with Down syn versus in ۰.۲% -۱.۴% of euploid fetuses and they also demonstrated that appropriate imaging of the nasal bone was attainable in a large percentage of the study population, with a ۹۹.۸% visualization rate.

- To assess the nasal bone sonographically, the fetal profile is first viewed in the midsagittal plane.
- The transducer can then be rocked sideways to maintain the angle of insonation at ۴۵ or ۱۳۵ degrees.
- The nasal bone is visualized as an echogenic line below and parallel to the overlying skin. When present, the nasal bone and skin appear similar to an equal sign.





Causes of Absent NB in first trimester

- In ۶۹% of cases of **trisomy** ۲۱,
- ۵۵% of trisomy ۱۸
- ۳۴% of trisomy ۱۳
- ۱۱% of monosomy X.

- In ۰.۶%–۲.۶% of the **euploid** population in the first trimester.

- *Varies by **crown-rump length***
- In ۴.۷% of euploid fetuses with a CRL of ۴۵–۵۴ mm
- ۳.۴% with CRL of ۵۵–۶۴ mm
- ۱.۴% with CRL of ۶۵–۷۴ mm
- ۱% with CRL of ۷۵–۸۴ mm.

- *Varies by **ethnicity***
- In ۲.۲% of whites
- ۳% of Chinese/ Japanese
- ۵.۰% of southern Asians
- ۹.۰% of Afro-Caribbeans.

- *Incidence of absent NB is **increased with wider nuchal translucency***
- ۱.۶% when NT is ≤ ۹۵%
- ۲.۷% when NT is ۹۵% or ۳.۴ mm
- ۵.۴% when NT is ۳.۵–۴.۴ mm
- ۶% when NT is ۴.۵–۵.۴ mm
- ۱۵% when NT is ≥۵.۵ mm

ANTENATAL SIGNIFICANCE

- **Combining** NT and serum biochemistry values results in a detection rate of **٩٠%** for Down syn ,**FPR with a of ٥.٠%.**
  Adding the NB evaluation decreases the **FPR to ٢.٥%** while maintaining a detection rate of **٩٠%.**
- Sensitivity of **absent NB alone** for detecting trisomy ٢١ is **٦٥%,** with a FPR of **٠.٨%.**

- The characteristics of the NB vary with **Gage**, NT **thickness**, and **ethnicity**, and these variations affect the **likelihood ratio**.

ANTENATAL SIGNIFICANCE

- **Absent of NB**  **Invasive Tests**

THE SECOND TRIMESTER GENETIC SONOGRAM

- The sonographic detection of a major structural abnormality significantly increases the risk that a fetal chromosome abnormality is present.
- Some structural abnormalities are particularly associated with a specific aneuploidy .
- However, many aneuploid fetuses, particularly those with Down syndrome, do not have major structural abnormalities that are readily detected in the first or second trimester.

- Because Down syndrome is the most common clinically significant chromosome abnormality, identification of minor features of Down syndrome, or so-called sonographic soft signs, is often employed as a screening tool.
- **One of most important soft markers is hypoplastic NB that also depends on etnicity.**
- As the association between ultrasound markers and Down syn became more apparent, the genetic sonogram was introduced as an alternative noninvasive method to further refine aneuploidy risk, most specifically for Down syn.

Likelihood ratio of Down syndrome based on the presence of an isolated soft marker (pooled results)

Finding	Sensitivity Down syndrome, percent	False positive rate (ie, marker detected in euploid karyotype), percent	Positive likelihood ratio if the marker is isolated, percent*
Absent or hypoplastic nasal bone	48.9 to 69.9	1.9 to 4.0	6.58
Aberrant right subclavian artery	17.9 to 47.4	1.0 to 2.1	3.94
Ventriculomegaly•	4.2 to 12.9	0.1 to 0.4	3.81
Increased nuchal fold△	20.3 to 32.9	0.5 to 1.9	3.79
Hyperechoic bowel◇	13.4 to 20.7	0.8 to 1.5	1.65
Pyelectasis§	11.2 to 17.2	1.4 to 2.0	1.08
Echogenic intracardiac focus	20.9 to 28.2	3.4 to 4.5	0.95
Short humerus	17.1 to 47.9	2.8 to 7.4	0.78
Short femur	19.3 to 38.1	4.7 to 8.8	0.61

The autnors concluded that if a systematic ultrasound examination is performed by expert sonologists and all of these markers are absent the risk of Down syndrome is the mother's a priori risk based on maternal serum screening multiplied by 0.13.

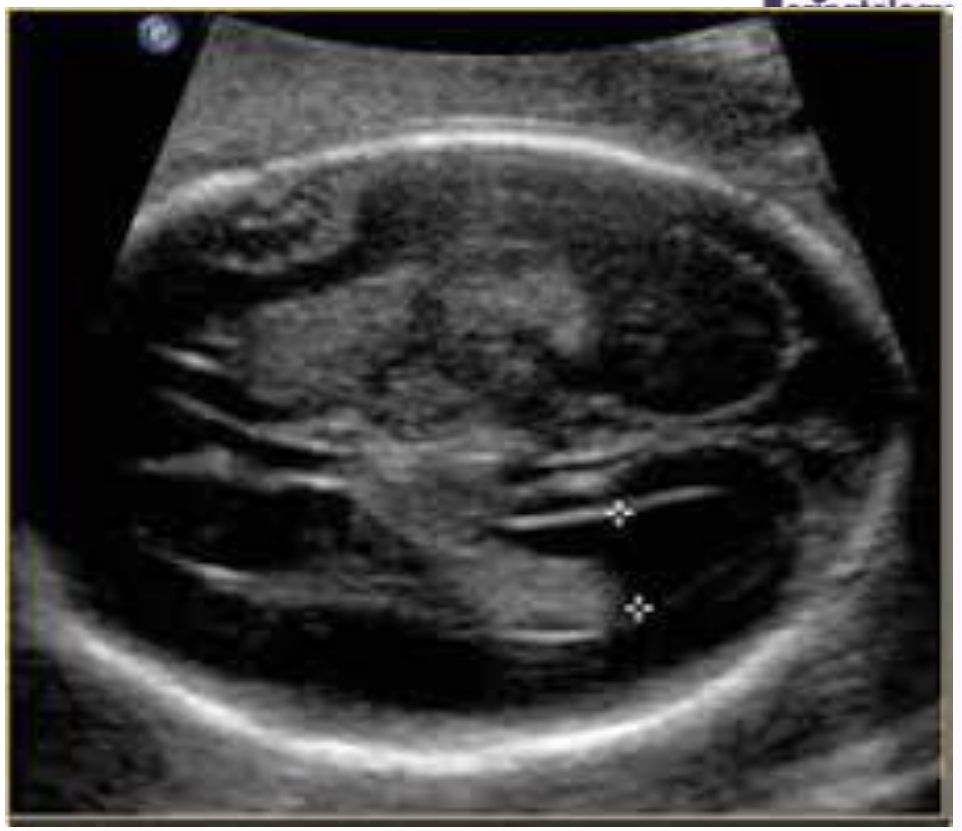
* Derived by multiplying the positive likelihood ratio for the marker by the negative likelihood ratio for each of the other markers.

http://perina

Fetal cerebral Ventriculomegaly

Definition

- Atrial diameter of LV between ۱۵ and ۴۰ wks remains stable, with reported means of ۵.۴ to ۷.۶ mm and an upper limit of normal of ۱۰ mm.
- Ventriculomegaly is mild if the atrial diameter is between ۱۰ - ۱۵ mm and severe if >۱۵ mm.
- Some authors use the categories of mild (۱۰ to ۱۲ mm), moderate (۱۳ to ۱۵ mm), and severe (≥۱۶ mm).
- Substantial interobserver variability in interpretation can occur, and is most common at borderline ventricular diameters



- Fetal cerebral ventriculomegaly is a relatively common finding on second trimester obstetrical ultrasound examination.

- It is clinically important because it can be caused by a variety of disorders that result in neurological, motor, and/or cognitive impairment.

Ventriculomegaly is "isolated" when the fetus has no other anomalies.

- Many cases that appear isolated prenatally are ultimately found to have other abnormalities, particularly when ventriculomegaly exceeds ۱۵ mm . These abnormalities include Chiari malformations, neural tube defects, Dandy Walker malformations, agenesis of the corpus callosum, and genetic syndromes.

- Hydrocephalus is the correct term for pathologic dilatation of the brain's ventricular system from increased pressure, usually due to obstruction.
- Ventriculomegaly is the appropriate term when dilatation is due to non obstructive causes, such as brain dysgenesis or atrophy.

- Most commonly, the term "ventriculomegaly" is used when the ventricles are mildly enlarged, and "hydrocephalus" is used when they measure >۱۵ mm.

PREVALENCE

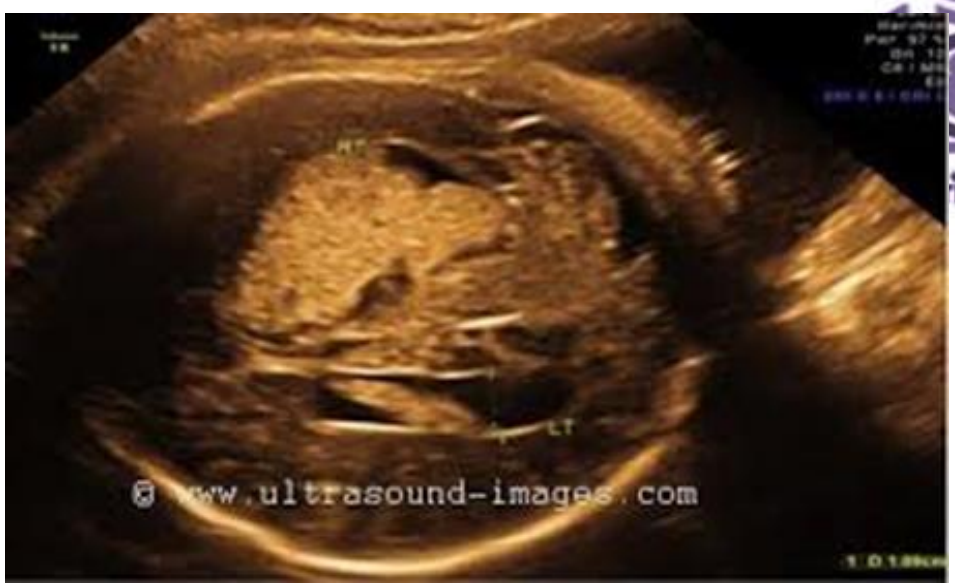
- Routine prenatal sonographic examinations between ۱۶ - ۲۲ (prevalence ۰.۱۵%.
- This is a reasonable assessment of the prevalence in a general obstetrical population undergoing second trimester prenatal sonography.
- Ventriculomegaly is more common in males; the male-to-female sex ratio is ۱.۷

ETIOLOGY

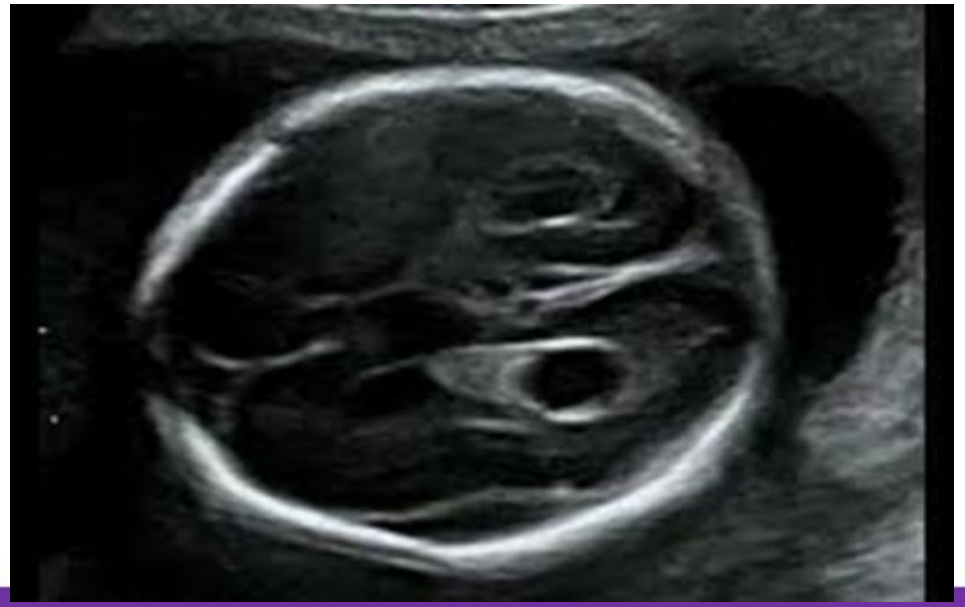
Pathologic causes of ventriculomegaly include:

- **Idiopathic** causes
- Chromosomal disorders (the most common is Tri ٢١)
- **Genetic** syndromes
- Congenital **infections** **TOXO,CMV,Zika**
- Aqueductal **stenosis**
- Cortical **malformations**
- **Migrational** abnormalities
- **Structural** abnormalities : corpus callosum agenesis, Dandy-Walker malformation, and neural tube defects,microcephally.

- Rarely, **overproduction of CSF** by a **tumor** or choroid plexus **papilloma** may result in ventriculomegaly.



- Large isolated **choroid plexus cysts** may transiently **dilate** the fetal cerebral ventricles. choroid plexus cysts are typically benign.



Significance

- In **mild to moderate** VM, chromosomal **aneuploidy** was seen in approximately **١١%.**
- Trisomy ٢١ was **most common**, followed by trisomies ١٨ and ١٣.

- **Structural anomalies** (with **normal karyotype**) were seen in **٤٣%** with **brain** anomalies being the **most common**, **heart**, **diaphragmatic hernia**, **omphalocele**, and **limb reduction**.

- Congenital **infection** was seen in ٠.٨% (CMV, toxo).

KEY DIAGNOSTIC FEATURES

- Lateral ventricle measuring ≥۱۰ mm but <۱۲ mm
- Choroid plexus may appear **dangling**.
- **Resolution** of mild isolated VM occurs in ۶۲% of cases < ۲۴ wks' gestation.
- **Males** more commonly have **mild** VM **than** females.

Management
ANTENATAL MONITORING

- Detailed anatomic survey

- Fetal neurologic scan to look for additional anomalies ,the head in the pelvis, transvaginal scan is helpful.

- Fetal echocardiogram

- Genetic counseling and amniocentesis, especially if mild VM is not an isolated finding or if maternal aneuploidy screening suggests risk.

- Consider sending **amniotic fluid** for infection studies (toxo and CMV for **PCR**).

Management
ANTENATAL MONITORING

- Zika virus if living in an area with active transmission
- MRI of the brain to look for additional brain anomalies
- Pediatric neurology consultation
- Serial ultrasound to monitor progression of VM

PROGNOSIS

- Fetuses with mild dilation (١٠ to ١٢ mm) and **no** other abnormalities have a normal postnatal evaluation in >٩٠%, suggesting this is a commonly normal variant.

- Even isolated moderate dilation (١٣ to ١٥ mm) has been associated with a normal postnatal examination in ٧٥ to ٩٣%.

PROGNOSIS

- In cases of mild VM associated with other anomalies, the prognosis is influenced by the anomaly (trisomy ۲۱ or agenesis of the corpus callosum).
- In utero progression of VM is associated with increased neurologic sequelae.
- In a review, the pooled prevalence of neurodevelopmental delay in isolated mild VM with normal karyotype was ۴.۹%
- ۱۰.۷% of females had neurodevelopment delay compared with ۵.۶% of males.

Thank You